

# Parents' Guide to Sickle Cell Disease

You have just learned that your infant has sickle cell disease (hemoglobin SS). Naturally you are concerned and have many questions. This information sheet will help answer some of your questions. However, it should not take the place of an informed discussion with your baby's doctor (primary care provider).

## What is Hemoglobin?

Hemoglobin is a protein in the red blood cells. It carries oxygen to all parts of the body and gives blood its red color. There are many hemoglobin types (this is not the same as a blood type). Hemoglobin is inherited through genes, one from each parent. Most people have hemoglobin A, also called adult hemoglobin. The presence of hemoglobin A makes the red blood cells smooth and round. These cells move easily through the blood vessels and deliver oxygen normally to the body.

## What is Sickle Cell Disease?

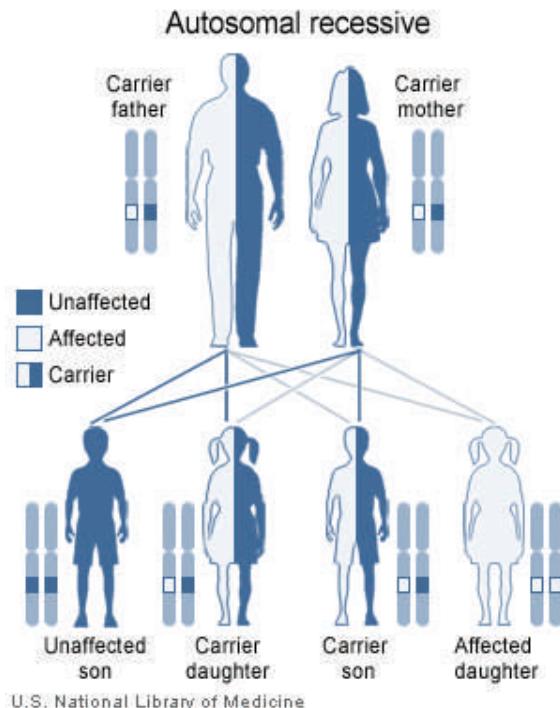
Hemoglobin S (sickle-shaped hemoglobin) in the red blood cells is responsible for causing sickle cell disease. Children *inherit* this disease from their parents as an recessive genetic disorder. This means a sickle cell (hemoglobin S) gene is passed from *both* mom and dad to the baby causing sickle cell disease (hemoglobin SS). When both parents have one hemoglobin S gene, there is a 1 in 4 or 25% chance with each pregnancy that an infant will inherit two hemoglobin S genes. Persons with sickle cell disease have periods of feeling well and episodes of sudden intense pain that can be anywhere in the body. This happens when sickled red blood cells become hard and sticky and clog up small blood vessels. Red blood cells cannot deliver oxygen to the body and over time may damage the body's tissues and organs. Sickle cell disease is not contagious.

## What is the Disease Frequency?

Sickle cell disease is found in males and females equally and occurs in all races. The highest frequency is among people of African, Mediterranean, Caribbean, Asian, Southeast Asian, and Central and South American descent. All newborn babies in Utah are tested for sickle cell disease, regardless of their race or ethnic background.

## What Problems can Sickle Cell Disease Cause ?

Children with sickle cell disease sometimes have serious health problems, if not treated, that can lead to death. These problems are: *infection of the blood* (septicemia) and *sudden enlargement of the spleen with a drop in the blood cell count* (acute splenic sequestration). Other serious problems include: sudden pain, swelling of



hands and feet, fever, increased infections, anemia, chest pain and trouble breathing, pneumonia, stroke, blood in urine, leg ulcers, gallstones, vision problems, yellow skin (jaundice), organ damage, kidney failure, painful erections, and problems during pregnancy.

## ***What can be done to Treat Sickle Cell Disease?***

If your child with sickle cell disease is having any of the symptoms listed above, parents should first consult the child's doctor for instructions.

■ **Pain Management.** Recommended home treatment includes applying a heating pad to the painful area and medicines such as acetaminophen (Tylenol®) or ibuprofen (Advil® and Motrin®). Drinking plenty of liquids and rest can help. Once the pain goes away children are usually active again. If the pain is not better after taking medicine at home, your child may need treatment with stronger medicine in the emergency room or to be admitted to the hospital for treatment.

■ **Penicillin.** Very serious infections of the blood sometimes occurs in infants and young children with sickle cell disease. Penicillin taken twice daily can prevent most of these infections.

## ***What are the Most Important Things to Remember ?***

- Work closely with your child's doctor and hematologist (a doctor who is a blood specialist). Make sure your child has regular checkups with them and call your child's doctor with questions..
- Your child should avoid extreme hot and cold temperatures and exhaustion, get plenty of rest, and drink lots of liquids to reduce his or her chances of having pain.
- Check your child's spleen as advised by the hematologist. Your child's doctor will show you where the spleen is and what feels normal. If the spleen suddenly feels larger, your child should be evaluated as soon as possible by his or her doctor.
- Your child will need an immediate medical evaluation for a fever of 38.5° C (101° F) or greater, difficulty breathing or chest pain. Take your child to a facility that provides emergency care.
- If your child has no energy and looks very pale he or she should be evaluated by a doctor.
- Be sure your child receives *all* childhood immunizations when they are scheduled and any additional immunizations recommended by your child's doctor.
- Call your child's doctor if you have questions and have your child seen if you have medical concerns.

## ***What Additional Treatment are Available?***

Blood transfusions and medications to decrease or prevent the formation of sickle-shaped red blood cells may be used. Sickle cell disease may be treated by a bone marrow transplant from a compatible donor, but is still considered a high-risk procedure.

## ***How Do I Get More Information?***

Talk with your baby's doctor. We recommend that you make an appointment with a pediatric hematologist in the near future. You may also want to have a genetic consultation for you and your family to see how sickle cell disease might affect future children or grandchildren.

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